A multi-omic approach to diagnosing rare disease

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The multi-omic approach to diagnosing rare disease

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INTRODUCING MULTI-OMICS

- Multi-omics is an approach to biological research which looks at multiple 'omes' including the **genomics**, **epigenomics** and **transcriptomics.**

(Check out the glossary below if you're not sure what any terms mean!)

- The 100,000 genomes project[2] is a UK based project which is attempting **whole genome sequencing** of patients with rare disease and their families to aid in diagnosis.
- From this data we can then carry out multi-omic research, increasing the likelihood of obtaining a diagnosis, which leads us to this project's research...

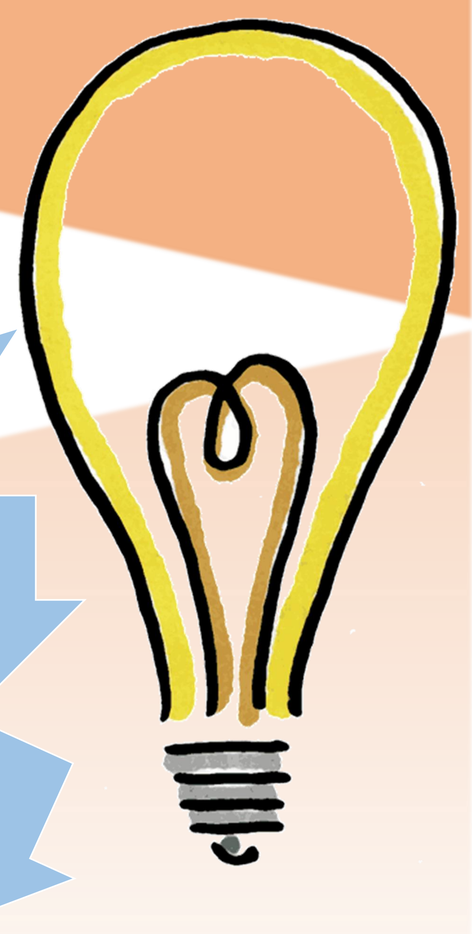
PATIENT CENTERED RESEARCH

- The paradox of rare disease is that whilst they are individually rare, approximately **350 million people worldwide**[1] are affected...
- There are an estimated **8000 types of rare disorders**[1], often with variations in clinical presentation, making diagnosis challenging...
- **Patients can wait several years for a diagnosis**, which can negatively impact prognosis, quality of life and make access to effective treatment and support difficult!

So what are researchers doing to help?

THIS RESEARCH PROJECT INVOLVES...

- Conducting **systematic reviews** into the current literature surrounding multi-omics and rare disease.
- Epigenomic and transcriptomic analyses of patient samples from the 100,000 genomes project which will have been subjected to whole genome sequencing but it was insufficient to render a diagnosis.
- Specifically I will be studying differential **methylation** (epigenomics) and **gene expression**, or **RNA levels** (transcriptomics).



Watch me!

Here's lots of patient experiences which give insights into living with a rare disease, including difficulties in getting a diagnosis.

HOPE FOR THE FUTURE

By utilising an allied multi-omic approach to studying rare disease we will be able to...

- Improve diagnosis speed and accuracy.
- Improve our understanding of the biological mechanisms behind rare disease.
- Identify potentially novel therapeutics.

Therefore, multi-omic approaches to studying rare disease will positively impact the lives of people living and working with rare disease.

Glossary of genetics terms

- **Genomics**: The study of the structure, function and evolution of a persons genetic material.
- **Epigenomics**: The study of non-sequence level DNA modifications which effects gene activity.
- **Methylation**: The act of adding a chemical methyl group which can effect gene activity levels.
- **RNA**: Ribonucleic acid which does many things, including acting as a messenger (mRNA) that carries instructions from the DNA to the cells to carry out cell duties, like synthesise proteins!
- **Gene expression**: The process of acting on the instructions contained in the active gene.
- **Transcriptomics**: The study of the total sum of mRNA which indicates gene expression levels.
- **Whole genome sequencing**: The impressive process of discovering the entire sequence of an organisms DNA in a single attempt, also known as high throughput sequencing.

QUEEN'S UNIVERSITY BELFAST

1. Global Genes. RARE Diseases: Facts and Statistics [Accessed 26/02/2018] Available from: https://globalgenes.org/rare-diseases-facts-statistics/
2. The 100,000 Genomes Project [Accessed 26/02/2018] Available from: https://www.genomicsengland.co.uk/the-100000-genomes-project/.